



ACVRL1 gene

activin A receptor like type 1

Normal Function

The *ACVRL1* gene provides instructions for making a protein called activin receptor-like kinase 1. This protein is found on the surface of cells, especially in the lining of developing arteries.

The ACVRL1 protein is a receptor. It acts as a "lock" waiting for a specific protein, called its ligand, to serve as the "key." In the case of the ACVRL1 protein, the ligand is called transforming growth factor beta. The interaction between these proteins plays a role in the development of blood vessels. In particular, this protein interaction is involved in the specialization of new blood vessels into arteries or veins.

Health Conditions Related to Genetic Changes

hereditary hemorrhagic telangiectasia

Dozens of mutations in the *ACVRL1* gene have been found to cause hereditary hemorrhagic telangiectasia type 2. Many *ACVRL1* gene mutations substitute one protein building block (amino acid) for another amino acid in the ACVRL1 protein, which impairs the protein's function. Other mutations prevent production of the ACVRL1 protein or result in an abnormally small protein that cannot function. The shortage of functional ACVRL1 protein appears to interfere with the development of boundaries between arteries and veins, resulting in the signs and symptoms of hereditary hemorrhagic telangiectasia type 2.

pulmonary arterial hypertension

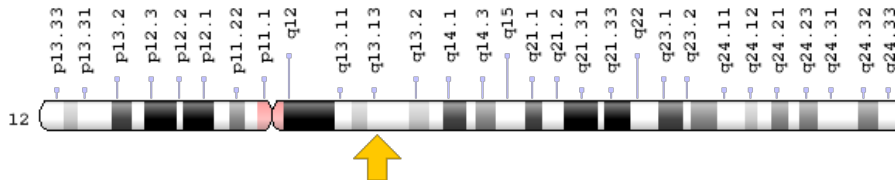
other disorders

A common genetic variation (polymorphism) in the *ACVRL1* gene has been found to appear more often in people who develop arteriovenous malformations in the brain, but who do not have other signs or symptoms of hereditary hemorrhagic telangiectasia, than in the general population. The polymorphism replaces a particular DNA building block (nucleotide) called adenine with the nucleotide guanine (written as IVS3-35 A>G). This genetic variation may affect the development of blood vessels in the brain, resulting in an increased risk of arteriovenous malformations.

Chromosomal Location

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 51,906,913 to 51,923,361 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- activin A receptor type II-like 1
- activin A receptor type II
- Activin A receptor, type II-like kinase 1
- ACVL1_HUMAN
- ACVRLK1
- ALK-1
- ALK1
- EC 2.7.1.37
- HHT
- HHT2
- ORW2
- Serine/threonine-protein kinase Receptor R3 Precursor
- SKR3
- TGF-B Superfamily Receptor Type I

Additional Information & Resources

GeneReviews

- Hereditary Hemorrhagic Telangiectasia
<https://www.ncbi.nlm.nih.gov/books/NBK1351>

Genetic Testing Registry

- GTR: Genetic tests for ACVRL1
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=94%5Bgeneid%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ACVRL1%5BTIAB%5D%29+OR+%28activin+A+receptor+type+II-like+1%5BTIAB%5D%29%29+OR+%28%28HHT%5BTIAB%5D%29+OR+%28ALK1%5BTIAB%5D%29+OR+%28HHT2%5BTIAB%5D%29+OR+%28ORW2%5BTIAB%5D%29+OR+%28SKR3%5BTIAB%5D%29+OR+%28ALK-1%5BTIAB%5D%29+OR+%28ACVRLK1%5BTIAB%5D%29+OR+%28Activin+A+receptor,+type+II-like+kinase+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- ACTIVIN A RECEPTOR, TYPE II-LIKE 1
<http://omim.org/entry/601284>
- ARTERIOVENOUS MALFORMATIONS OF THE BRAIN
<http://omim.org/entry/108010>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/ACVRL1ID569ch12q13.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ACVRL1%5Bgene%5D>
- HGNC Gene Family: Type 1 receptor serine/threonine kinases
<http://www.genenames.org/cgi-bin/genefamilies/set/345>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=175
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/94>
- UniProt
<http://www.uniprot.org/uniprot/P37023>

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